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Case report

Rare Prenatal Association of complete Trisomy 15 with 69, XXX Triploidy: Case Report and Literature Review

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Abstract

Background: Trisomy 15 and triploidy are both rare chromosomal abnormalities usually observed in early pregnancy losses. Their coexistence in a viable gestation has never been reported. This case describes an exceptional prenatal association of complete trisomy 15 and 69XXX triploidy, providing detailed clinical, sonographic, and cytogenetic documentation.

Case presentation: A 41-year-old pregnant woman underwent a second-trimester morphology ultrasound showing severe, proportionate fetal growth restriction, absent nasal bones, and clenched hands. Amniocentesis at 26 weeks identified 69, XXX by FISH, and conventional karyotyping subsequently revealed complete trisomy 15. Multidisciplinary counseling led to termination at 29 weeks. Postmortem examination confirmed a female phenotype with craniofacial dysmorphia (flat forehead, mild nasal flaring, hypertelorism, retrognathism), elongated limbs with clenched hands, and a bulging thorax, without visceral or cerebral malformations.

Conclusions: This case illustrates an exceptionally uncommon combination of numerical chromosomal abnormalities. Although isolated trisomy 15 and triploidy can each present with growth restriction and multiple abnormalities, their co-occurrence likely confers near-certain lethality. Early detailed ultrasound followed by definitive cytogenetic testing is critical for counseling and management in suspected multiple aneuploidies.

Keywords: trisomy 15; 69XXX triploidy; multiple aneuploidy; prenatal diagnosis; fetopathology; fetal growth restriction

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1. Introduction

Trisomy 15 and triploidy are both rare chromosomal abnormalities, most often identified in the context of spontaneous abortions or severe prenatal malformations [1]. The estimated incidence of complete trisomy 15 is extremely low, accounting for approximately 1% of chromosomal anomalies in early pregnancy losses, whereas triploidy occurs in about 1-2 % of conceptions [2]. In ongoing pregnancies, both entities are typically associated with profound growth restriction, multiple structural anomalies, and placental abnormalities [3]. This report provides detailed clinical, sonographic, and cytogenetic documentation of these double aneuploidies and discusses potential underlying mechanisms, phenotypic features, and implications for genetic counseling.

2. Case Presentation

A 41-year-old pregnant woman, gravida 2 para 1, with no history of consanguinity or genetic disease, was referred for a second-trimester morphology ultrasound for detailed ultrasound evaluation. The maternal medical history was unremarkable.

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Ultrasound finding

A detailed fetal ultrasound revealed severe intrauterine growth restriction with biometric parameters below the third percentile for gestational age. The biparietal diameter, head circumference, and abdominal circumference corresponded to approximately -3SD. A second-trimester ultrasound has demonstrated agenesis of the nasal bones and clenched hands (Figure 1).

Cytogenetic analysis

An amniocentesis was performed at 26 weeks. Conventional karyotyping revealed a “69, XXX triploidy” pattern, while interphase fluorescence in situ hybridization using centromeric probes for chromosomes 13,18,21, X, and Y confirmed triploidy. Further extended analysis identified a concomitant complete trisomy 15.

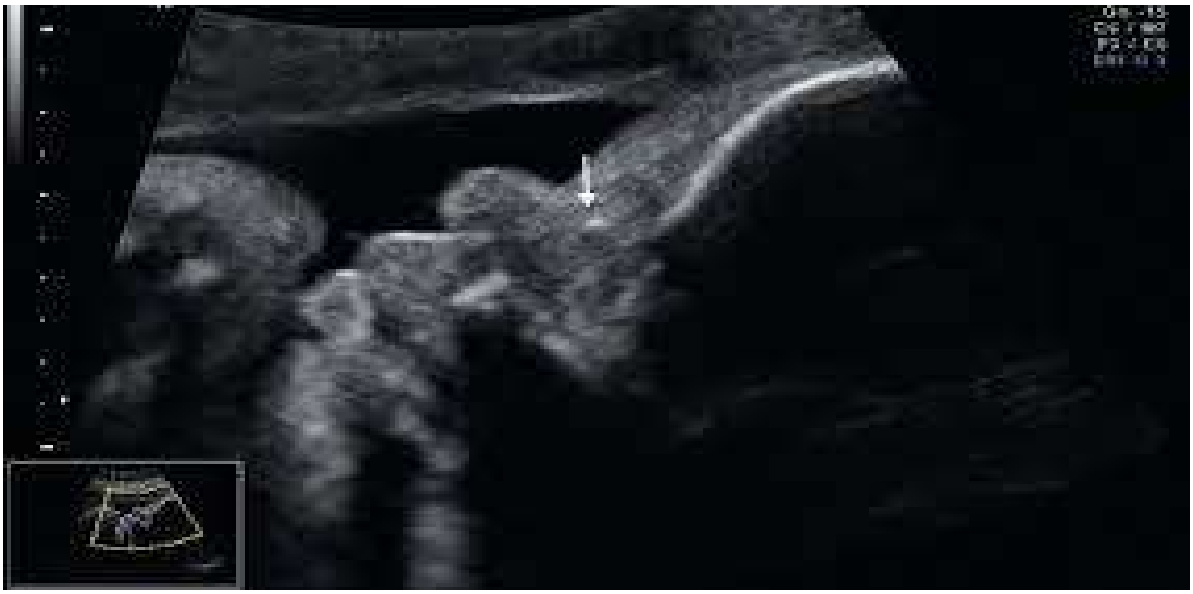

Fig 1. Strict sagittal section of the fetal profile at 23 weeks of gestation showing agenesis of the nasal bones.

Pathological findings

After multidisciplinary counseling and informed consent, termination of pregnancy was undertaken at 29 weeks. Fetal examination showed a female phenotype with distinctive craniofacial dysmorphia, long neck, flat forehead, mild nasal flaring, hypertelorism, retrognathism, elongated limbs with clenched hands, and a bulging thorax. No visceral or brain malformations were found on organ dissection, and the neuroanatomic examination showed no structural anomalies (Figure 2).

3. Discussion

Cytogenetic interpretation and mechanisms

The coexistence of a complete trisomy 15 with another chromosomal abnormality represents an exceptional event, reflecting complex errors occurring during gametogenesis or early embryogenesis [4]. Several mechanisms may account for this observation.

Fig 2: Coronal facial view demonstrating hypertelorism and a short nasal bone.

Dispermy, where two spermatozoa fertilize a single oocyte, may cause a triploid chromosomal complement with maternal and paternal sets. In our case, the coexistence of complete trisomy15 and triploidy likely reflects a rare meiotic or postzygotic event involving nondisjunction and dispermy. Alternatively, retention of a polar body or failure of maternal meiosis I or II segregation can result in an additional maternal chromosome 15. Postzygotic mechanisms such as mitotic nondisjunction or trisomy rescue leading to uniparental disomy (UPD) may also contribute to the chromosomal imbalance. Determining the parental origin of the extra chromosome through molecular genotyping (microsatellite or SNP analysis) would help clarify the underlying mechanism and guide recurrence risk assessment in future pregnancies [5].

Phenotype and comparison with the literature

The phenotype of complete trisomy 15 is rarely documented because of its nearly universal lethality. Reported cases consistently describe severe intrauterine growth restriction (IUGR), craniofacial dysmorphism, and multiple internal malformations, particularly affecting the cardiovascular and central nervous systems [5,6]. These manifestations overlap with those observed in triploid conceptuses, suggesting that both chromosomal abnormalities interfere with early embryonic development through shared pathogenic pathways. In the present case, the association of profound growth restriction, facial anomalies, and placental hydropic changes is consistent with previously published descriptions. Compared with isolated trisomy 15, our case exhibited more pronounced placental abnormalities and no visceral malformations, supporting a possible synergistic deleterious effect of the double aneuploidy [7]. Similar findings have been reported in other combined aneuploidies, such as trisomy 13 with triploidy or trisomy 18 with 45, X mosaicism, which also demonstrate increased developmental disruption and early lethality [8].

Genetic counseling and recurrence risk

From a counseling perspective, the risk of recurrence for full trisomy 15 associated with triploidy is considered extremely low and usually sporadic. However, parental karyotyping remains recommended to exclude balanced rearrangements or germline mosaicism that may predispose to recurrent meiotic errors [9,10]. When no parental chromosomal abnormality is detected, the event can be considered *de novo*. The prognosis of complete trisomy 15, whether isolated or associated with another aneuploidy, remains uniformly fatal due to early developmental arrest and severe malformations [11]. Future pregnancies should be monitored with first-trimester combined screening and non-invasive prenatal testing (NIPT), followed by early targeted ultrasonography in the presence of suspicious findings [12,13]. Comprehensive cytogenetic and pathological evaluation of such exceptional cases contributes to the refinement of diagnostic criteria and enhances understanding of chromosomal mechanisms in early embryonic lethality [14]. This case represents an exceptionally rare coexistence of complete trisomy 15 and triploidy, contributing new insights into prenatal cytogenetic abnormalities.

Conclusion

This case highlights the importance of comprehensive ultrasound surveillance and cytogenetic analysis, with precise reporting to elucidate pathogenic mechanisms and recurrence risk [15]. Given the diagnostic and counseling complexities, such cases should prompt multidisciplinary management, including genetic counseling with discussion of targeted testing in subsequent pregnancies [16]. It adds to the limited body of literature on double aneuploidies and underscores the need for careful prenatal evaluation.

Statements and Declarations

Ethics approval and consent

Institutional ethical standards were followed. Written informed consent for anonymized reporting and image use was obtained from the parents.

Competing interests

The authors declare no competing interests.

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No funding was received for this work.

Data availability

All data relevant to the case are included in the article. Additional details are available from the corresponding author upon reasonable request.

Authors' contributions

RB and DB managed the case and drafted the manuscript. DBa, AH, SBS, and AL contributed to data acquisition, fetopathology interpretation, and critical revisions. All authors approved the final manuscript and agreed to be accountable for all aspects of the work.

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